

DIGITAL KARYOTYPING

Abstract of the Invention

Alterations in the genetic content of a cell underlie many human diseases, including cancers. A method called Digital Karyotyping provides quantitative analysis of DNA copy number at high resolution. This approach involves the isolation and enumeration of short sequence tags from specific genomic loci. Analysis of human cancer cells using this method identified gross chromosomal changes as well as amplifications and deletions, including regions not previously known to be altered. Foreign DNA sequences not present in the normal human genome could also be readily identified. Digital Karyotyping provides a broadly applicable means for systematic detection of DNA copy number changes on a genomic scale.